Researchers find new genetic pathway behind neurodevelopmental disorders

December 6 2012

Researchers at the Douglas Mental Health University Institute, have discovered a new genetic process that could one day provide a novel target for the treatment of neurodevelopmental disorders, such as intellectual disability and autism.

The research study, which appears in the December issue of the American Journal of Human Genetics, was led by Carl Ernst, a Douglas Institute researcher, an assistant professor in McGill's Department of Psychiatry and a Canada Research Chair in Psychiatric Genetics. Ernst and his colleagues found that genetic mutations that negatively affect brain development can occur in a gene family of previously unknown function in the human genome.

According to the World Health Organization, neurodevelopmental disorders affect one in six children in industrialized countries. Impairing the growth and development of the brain or central nervous system, neurodevelopmental disorders encompass a broad range of conditions, including developmental delay, autism spectrum disorders and cerebral palsy. People with neurodevelopmental disorders can experience difficulties with language, speech, learning, behaviour, motor skills and memory.

Mutations in genes are thought to underlie many neurodevelopmental disorders, but all genes important for brain development found to date are in a single pathway. Genes are coded in DNA that gives way to RNA, which gives way to protein. Proteins form the functional unit of the body

and are the major players in all biological activity. Prior to the current study, all genetic mutations important for neurodevelopmental disorders, occured in genes that make protein.

The work of Ernst and his research team identified an important shortcut in the process of making functional molecules for brain development. By sequencing the genomes of 200 people with neurodevelopmental disorders and chromosomal abnormalities, and comparing the results to more than 15,000 control samples, the researchers made a surprising discovery: some individuals had mutations in a gene that did not make protein.

"Our discovery tells us that mutations in genes that code only for RNA and do not make protein can have a functional impact and lead to neurodevelopmental abnormalities," Ernst says. "In previous studies of brain development, RNA was just considered a middle player – one that only served as a template for the production of proteins."

By opening up a new area of study involving RNA, Ernst aims to advance understanding of the underlying causes of neurodevelopmental disorders. "We hope to shine a new light on how the brain develops," he says.

Provided by Douglas Mental Health University Institute

Citation: Researchers find new genetic pathway behind neurodevelopmental disorders (2012, December 6) retrieved 10 April 2024 from https://medicalxpress.com/news/2012-12-genetic-pathway-neurodevelopmental-disorders.html

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